



Genetics Home Reference

Your Guide to Understanding Genetic Conditions

Help Me Understand Genetics

Chapter 4

Genetic Testing

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Chapter 4

Genetic Testing

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What is genetic testing?

Genetic testing is a type of medical test that identifies changes in chromosomes, genes, or proteins. Most of the time, testing is used to find changes that are associated with inherited disorders. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder. Several hundred genetic tests are currently in use, and more are being developed.

Genetic testing is voluntary. Because testing has both benefits and limitations, the decision about whether to be tested is a personal and complex one. A genetic counselor can help by providing information about the pros and cons of the test and discussing the social and emotional aspects of testing.

For general information about genetic testing:

MedlinePlus offers a list of links to [information about genetic testing \(http://www.nlm.nih.gov/medlineplus/genetictesting.html\)](http://www.nlm.nih.gov/medlineplus/genetictesting.html).

The National Human Genome Research Institute provides an overview of this topic as part of its [Frequently Asked Questions About Genetics \(http://www.genome.gov/10001191#3\)](http://www.genome.gov/10001191#3).

The Genetics and Public Policy Center also offers [information about genetic testing \(http://www.dnapolicy.org/genetics/testing.jhtml\)](http://www.dnapolicy.org/genetics/testing.jhtml).

What are the uses of genetic testing?

Genetic testing can provide information about a person's genes and chromosomes throughout life. Available types of testing include:

Newborn screening

Newborn screening is used just after birth to identify genetic disorders that can be treated early in life. The routine testing of infants for certain disorders is the most widespread use of genetic testing—millions of babies are tested each year in the United States. All states currently test infants for phenylketonuria (a genetic disorder that causes mental retardation if left untreated) and hypothyroidism (a disorder of the thyroid gland). Some states also test for other genetic disorders.

Diagnostic testing

Diagnostic testing is used to diagnose or rule out a particular genetic or chromosomal condition. It is usually offered to people who have signs of a particular disorder. This type of testing can be performed at any time during a person's life, but is not available for all genetic conditions. The results of a diagnostic test can influence a person's choices about health care and the management of symptoms.

Carrier testing

Carrier testing is used to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder. This type of testing is offered to individuals who have a family history of a genetic disorder and to people in ethnic groups with an increased risk of specific genetic conditions. If both parents are tested, the test can provide information about a couple's risk of having a child with a genetic condition.

Prenatal testing

Prenatal testing is used to detect changes in a fetus's genes or chromosomes before birth. This type of testing is offered to couples with an increased risk of having a baby with a genetic or chromosomal disorder. In some cases, prenatal testing can lessen a couple's uncertainty or help them make decisions about a pregnancy. It cannot identify all possible inherited disorders and birth defects, however.

Predictive and presymptomatic testing

These types of testing are used to detect gene mutations associated with disorders that appear later in life. Predictive testing can identify mutations that increase a person's risk of developing disorders with a genetic basis, such as certain types of cancer. Presymptomatic testing can determine whether a person will develop a genetic disorder, such as Huntington disease (an inherited brain disorder that appears during mid-life), before any symptoms appear. The results of predictive and presymptomatic testing can help people make decisions about medical care.

Forensic testing

Forensic testing uses DNA sequences to identify an individual for legal purposes. Unlike the tests described above, forensic testing is not used to detect gene mutations associated with disease. This type of testing can identify crime or catastrophe victims, rule out or implicate a crime suspect, or establish biological relationships between people (for example, paternity).

For more information about the uses of genetic testing:

Information about the types of genetic testing is available from [GeneTests](http://www.genetests.org/) (<http://www.genetests.org/>). Click on “Educational Materials” at the top of the GeneTests home page and scroll down to “Uses of Genetic Testing.”

The [National Newborn Screening and Genetics Resource Center](http://genes-r-us.uthscsa.edu/) (<http://genes-r-us.uthscsa.edu/>) offers detailed information about newborn screening.

For information about forensic DNA testing, refer to the fact sheet [DNA Forensics](http://www.ornl.gov/TechResources/Human_Genome/elsi/forensics.html) (http://www.ornl.gov/TechResources/Human_Genome/elsi/forensics.html) from the U.S. Department of Energy Office of Science.

How is genetic testing done?

Once a person decides to proceed with genetic testing, a medical geneticist, genetic counselor, primary care doctor, or specialist can order the test. Genetic testing is often done as part of a genetic consultation.

Genetic tests are performed on a sample of blood, hair, skin, amniotic fluid (the fluid that surrounds a fetus during pregnancy), or other tissue. For example, a procedure called a buccal smear uses a small brush or cotton swab to collect a sample of cells from the inside surface of the cheek. The sample is sent to a laboratory where technicians look for specific changes in chromosomes, DNA, or proteins, depending on the suspected disorder. The laboratory reports the test results in writing to a person's doctor or genetic counselor.

Newborn screening tests are done on a small blood sample, which is taken by pricking the baby's heel. Unlike other types of genetic testing, a parent will usually only receive the result if it is positive. If the test result is positive, additional testing is needed to determine whether the baby has a genetic disorder.

Before a person has a genetic test, it is important that he or she understands the testing procedure, the benefits and limitations of the test, and the possible consequences of the test results. The process of educating a person about the test and obtaining permission is called informed consent.

For more information about genetic testing procedures:

[GeneTests \(http://www.genetests.org/\)](http://www.genetests.org/) explains the testing process and informed consent. Click on “Educational Materials” at the top of the GeneTests home page and scroll down to “Ordering Genetic Testing.”

For information about how newborn screening tests are performed, refer to [Genetic Testing of Newborn Infants \(http://gslc.genetics.utah.edu/units/newborn/samples.cfm\)](http://gslc.genetics.utah.edu/units/newborn/samples.cfm) from the University of Utah Genetic Science Learning Center.

What is the cost of genetic testing, and how long does it take to get the results?

The cost of genetic testing can range from under \$100 to more than \$2,000, depending on the nature and complexity of the test. The cost increases if more than one test is necessary or if multiple family members must be tested to obtain a meaningful result. For newborn screening, costs vary by state. Some states cover part of the total cost, but most charge a fee of \$15 to \$60 per infant.

From the date that a sample is taken, it may take a few weeks to several months to receive the test results. Results for prenatal testing are usually available more quickly because time is an important consideration in making decisions about a pregnancy. The doctor or genetic counselor who orders a particular test can provide specific information about the cost and time frame associated with that test.

For more information about the costs and turnaround time for genetic tests:

[GeneTests \(http://www.genetests.org/\)](http://www.genetests.org/) provides a list of factors that influence the turnaround time and costs of genetic testing. Click on “Educational Materials” at the top of the GeneTests home page and scroll down to “Ordering Genetic Testing.” In this section, “Turn-Around Time” and “Cost” are found under the heading “Choosing a Laboratory.”

Will health insurance cover the costs of genetic testing?

In many cases, health insurance plans will cover the costs of genetic testing when it is recommended by a person's doctor. Health insurance providers have different policies about which tests are covered, however. A person interested in submitting the costs of testing may wish to contact his or her insurance company beforehand to ask about coverage.

Some people may choose not to use their insurance to pay for testing because the results of a genetic test can affect a person's health insurance coverage. Instead, they may opt to pay out-of-pocket for the test. People considering genetic testing may want to find out more about their state's privacy protection laws before they ask their insurance company to cover the costs. (Refer to the question “What is genetic discrimination?” for more information.)

What are the benefits of genetic testing?

Genetic testing has potential benefits whether the results are positive or negative for a gene mutation. Test results can provide a sense of relief from uncertainty and help people make informed decisions about managing their health care. For example, a negative result can eliminate the need for unnecessary checkups and screening tests in some cases. A positive result can direct a person toward available prevention, monitoring, and treatment options. Some test results can also help people make decisions about having children. Newborn screening can identify genetic disorders early in life so treatment can be started as early as possible.

For more information about the benefits of genetic testing:

The National Cancer Institute provides a brief discussion of the [benefits of genetic testing](http://press2.nci.nih.gov/sciencebehind/genetesting/genetesting30.htm) (<http://press2.nci.nih.gov/sciencebehind/genetesting/genetesting30.htm>).

Additional information on this topic is available in the fact sheet [Gene Testing](http://www.ornl.gov/TechResources/Human_Genome/medicine/genetest.html#procon) (http://www.ornl.gov/TechResources/Human_Genome/medicine/genetest.html#procon) from the U.S. Department of Energy Office of Science.

What are the risks and limitations of genetic testing?

The physical risks associated with most genetic tests are very small, particularly for those tests that require only a blood sample or buccal smear (a procedure that samples cells from the inside surface of the cheek). The procedures used for prenatal testing carry a small but real risk of losing the pregnancy (miscarriage) because they require a sample of amniotic fluid or tissue from around the fetus.

Many of the risks associated with genetic testing involve the emotional, social, or financial consequences of the test results. People may feel angry, depressed, anxious, or guilty about their results. In some cases, genetic testing creates tension within a family because the results can reveal information about other family members in addition to the person who is tested. The possibility of genetic discrimination in employment or insurance is also a concern. (Refer to the question “What is genetic discrimination?” for additional information.)

Genetic testing can provide only limited information about an inherited condition. The test often can't determine if a person will show symptoms of a disorder, how severe the symptoms will be, or whether the disorder will progress over time. Another major limitation is the lack of treatment strategies for many genetic disorders once they are diagnosed.

A genetics professional can explain in detail the benefits, risks, and limitations of a particular test. It is important that any person who is considering genetic testing understand and weigh these factors before making a decision.

For more information about the risks and limitations of genetic testing:

The National Cancer Institute provides a brief discussion of the limitations of genetic testing:

[Limitations of Gene Testing \(http://press2.nci.nih.gov/sciencebehind/genetesting/genetesting31.htm\)](http://press2.nci.nih.gov/sciencebehind/genetesting/genetesting31.htm)

[Major Limitations of Gene Testing \(http://press2.nci.nih.gov/sciencebehind/genetesting/genetesting32.htm\)](http://press2.nci.nih.gov/sciencebehind/genetesting/genetesting32.htm)

Additional information about the risks and benefits of genetic testing can be found in the publication [Genomics and Its Impact on Science and Society: The Human Genome Project and Beyond \(http://www.ornl.gov/TechResources/Human_Genome/publicat/primer2001/6.html\)](http://www.ornl.gov/TechResources/Human_Genome/publicat/primer2001/6.html) from the U.S. Department of Energy Office of Science. Scroll down to the section “Gene Testing.”

GeneTests (<http://www.genetests.org/>) outlines points to consider for each type of genetic testing. Click on “Educational Materials” at the top of the GeneTests home page and scroll down to “Uses of Genetic Testing.”

What is genetic discrimination?

Genetic discrimination occurs when people are treated differently by their employer or insurance company because they have a gene mutation that causes or increases the risk of an inherited disorder. People who undergo genetic testing may be at risk for genetic discrimination.

The results of a genetic test are normally included in a person's medical records. When a person applies for life, disability, or health insurance, the insurance company may ask to look at these records before making a decision about coverage. An employer may also have the right to look at an employee's medical records. As a result, genetic test results could affect a person's insurance coverage or employment. People making decisions about genetic testing should be aware that when test results are placed in their medical records, the results might not be kept private.

Fear of discrimination is a common concern among people considering genetic testing. Several laws at the federal and state levels help protect people against genetic discrimination; however, genetic testing is a fast-growing field and these laws don't cover every situation.

For more information about privacy and genetic discrimination:

The National Human Genome Research Institute provides a detailed discussion of genetic discrimination and current laws that address this issue:

[Health Insurance and Employment Discrimination \(http://www.genome.gov/10002347\)](http://www.genome.gov/10002347)

[Privacy and Discrimination in Genetics \(http://www.genome.gov/10002077\)](http://www.genome.gov/10002077)

The Genetic Alliance offers links to resources and policy statements on [genetic discrimination \(http://www.geneticalliance.org/geneticissues/discrimresources.html\)](http://www.geneticalliance.org/geneticissues/discrimresources.html).

Additional information about [policy and legislation related to genetic privacy \(http://www.ornl.gov/TechResources/Human_Genome/elsi/legislat.html\)](http://www.ornl.gov/TechResources/Human_Genome/elsi/legislat.html) is available from the U.S. Department of Energy Office of Science.

How does genetic testing in a research setting differ from clinical genetic testing?

The main differences between clinical genetic testing and research testing are the purpose of the test and who receives the results. The goals of research testing include finding unknown genes, learning how genes work, and advancing our understanding of genetic conditions. The results of testing done as part of a research study are usually not available to patients or their healthcare providers. Clinical testing, on the other hand, is done to find out about an inherited disorder in an individual patient or family. People receive the results of a clinical test and can use them to help them make decisions about medical care or reproductive issues.

It is important for people considering genetic testing to know whether the test is available on a clinical or research basis. Clinical and research testing both involve a process of informed consent in which patients learn about the testing procedure, the risks and benefits of the test, and the potential consequences of testing.

For more information about the differences between clinical and research testing:

GeneTests (<http://www.genetests.org/>) outlines the major differences between clinical tests and research tests. Click on “Educational Materials” at the top of the GeneTests home page and scroll down to the question “What is Genetic Testing?”

The Genetic Alliance fact sheet **Informed Consent: Participation in Genetic Research Studies** (<http://www.geneticalliance.org/geneticissues/informedconsent.html>) provides information about informed consent and questions to ask genetic researchers.



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